



# GENETICS BRIEF

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## Chronic Disease and Genetics

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As the science of genetics has matured, research has shifted focus from rare, single gene disorders to common, multifactorial chronic diseases. Chronic disease affects more than 90 million Americans, accounting for 70% of all deaths and 60% of the nation's medical costs. As research progresses, genetics offers the opportunity to target health promotion and disease prevention programs better and the possibility to conserve health-care program resources. However, the contribution of genetics to chronic disease is complex, complicated by the interaction of genes with the environment, and with one another. Here we examine the role of genetics in five chronic diseases: cardiovascular disease, diabetes, and breast, colon and prostate cancer.

### Cardiovascular Disease

According to the Centers for Disease Control and Prevention, cardiovascular disease, principally heart disease and stroke, is the leading cause of death among men and women, across all racial and ethnic groups. Cardiovascular disease affects approximately 58 million Americans and costs the nation \$274 billion each year, including health expenditures and lost productivity. Research has begun to uncover a number of potential genetic susceptibility genes for heart disease and stroke and their risk factors (e.g., obesity and high blood pressure).

**Heart Disease.** Heart disease has become a major focus of genetic research. In the last decade the number of publications on genetic contributions to heart disease has risen exponentially. Genetic mutations have been associated with various risk factors for heart disease, including lipid metabolism and transport, high blood pressure, and elevated blood plasma homocysteine levels. It is believed that while traditional risk factors including environmental influences explain approximately 50% of cardiovascular disease, genetics may help explain the remaining disease burden.

**Stroke.** Studies also have indicated a genetic predisposition to both ischemic and hemorrhagic stroke. Although single gene disorders explain a small fraction of strokes, the genetic contribution to stroke most likely will be multifactorial and complex. Recent family studies, including the Framingham Heart Study, have found a significant genetic component to stroke.

### Diabetes

Diabetes, characterized by either a shortage of insulin or a decreased ability to use insulin, is a serious and increasingly common disease. It affects almost 16 million Americans and costs nearly \$100 billion a year. Diabetes is the seventh leading cause of death in the United States, and has many complications, including blindness, kidney failure, amputations, pregnancy complications, and increased risk of death due to influenza and pneumonia.

Genetic risk factors have been identified for type I and II diabetes. Type I diabetes has been associated with the mutations on chromosome 6 and chromosome 11. Various mutations in multiple genes have been linked to type II diabetes, with each group of mutations most likely contributing a small percentage to the overall

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risk. A few variations of type I and II diabetes have been found to be linked to mutations occurring in a single gene, however, most cases of the disease are likely to involve more than one gene.

## Cancer

According to Steven S. Coughlin and Wylie Burke, of the CDC and University of Washington, respectively, “the identification and the evaluation of inherited risk are likely to play an increasingly important role in cancer prevention strategies” (Burke, W. and S. Coughlin, 2000). Using genetic medicine to target health promotion better and thereby reduce modifiable risk factors for people with an inherited predisposition for cancer represents a new paradigm for cancer prevention.

**Breast Cancer.** Breast cancer is the most common cancer among women, and the second leading cause of cancer-related death for women. The BRCA1 and BRCA2 mutations account for approximately 5% of breast cancers, and up to 10% of ovarian cancers. One to two people per 1,000 carry these mutations, which lead to elevated risks of breast and ovarian cancer. However, it is not known what percentage of mutation carriers will develop the disease, a concept known as penetrance. If a condition is expressed in less than 100 percent of persons who carry the specific mutation, as is the case with BRCA 1 or 2, then the penetrance is said to be less than 100 percent or the penetrance is reduced. Other potential genetic risk factors are being studied, including genes that are likely to cause disease onset. There is hope that genetic advances will result in preventive and treatment interventions for at-risk populations.

**Colon Cancer.** Colon cancer is one of the most common cancers in the U.S. population, and the second leading cause of cancer death. Several susceptibility genes have been identified for the inherited syndromes familial adenomatous polyposis (FAP) and hereditary nonpolyposis colorectal cancer (HNPCC). FAP, which occurs in 1 in 10,000 people, is caused by mutations on chromosome 5. HNPCC occurs in every 200-400 people in the U.S. and is one of the most common inherited diseases. As with other chronic diseases, it is believed that genetic testing for colon cancer may offer opportunities for improved health promotion and disease prevention and treatment strategies.

**Prostate Cancer.** Prostate cancer is the second most commonly diagnosed cancer in the United States, and is the second leading cause of cancer death for men. A possible genetic susceptibility locus has been found, but no specific genes have been located. The genetic contribution to prostate cancer is complex, and there is a need for knowledge of the molecular genetics of prostate cancer.

## Summary

Genetics will play an important role in health promotion and treatment and prevention strategies for chronic diseases. There is a need for risk communication to the public regarding the meaning of genetic discovery and health status. Translational research that takes the discovery of disease susceptibility genes and creates opportunities for better-targeted treatment and prevention strategies is imperative to the development of better prevention and treatment interventions for chronic diseases.

## Selected References

Coughlin, S.S. and W. Burke. “Public Health assessment of genetic predisposition to cancer.” *Genetics and Public Health in the 21<sup>st</sup> Century*. Eds. Khoury, M.J., Burke, W., and E.J. Thomson. Oxford University Press, 2000. pp. 151-171.

## Resources

Centers for Disease Control and Prevention National Center for Chronic Disease Prevention and Health Promotion. <http://www.cdc.gov/nccdphp>.

Centers for Disease Control and Prevention Office of Genetics and Disease Prevention. <http://www.cdc.gov/genetics>.

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